

# Distribution of Y-chromosomal haplotypes in the Basque Country autochthonous population using a 17-locus multiplex PCR assay

Oscar García<sup>a,\*</sup>, Iñaki Yurrebaso<sup>a</sup>, Ion Uriarte<sup>a</sup>,  
Juan Antonio Pérez<sup>a</sup>, Raúl Peñas<sup>a</sup>, Santos Alonso<sup>b</sup>,  
Concepción de la Rúa<sup>b</sup>, Neskuts Izagirre<sup>b</sup>, Carlos Flores<sup>c</sup>,  
Pablo Martín<sup>d</sup>, Cristina Albarrán<sup>d</sup>, Antonio Alonso<sup>d</sup>

<sup>a</sup> *Area de Laboratorio Ertzaintza, Sección de Genética Forense, Erandio (Bizkaia), Basque Country, Spain*

<sup>b</sup> *Depart. Genética, Antropología Física y Fisiología Animal, Universidad País Vasco, Bilbao, Basque Country, Spain*

<sup>c</sup> *Unidad Investigación, Hosp. Univ. “Nuestra Señora de Candelaria”, Servicio Canario Salud, Tenerife, Spain*

<sup>d</sup> *Instituto de Toxicología y Ciencias Forenses, Sección de Biología, Madrid, Spain*

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**Abstract.** Haplotype, allele frequencies and population data of 17 Y-chromosome STR loci DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 (Y GATA C4) and Y GATA H4 were determined from a sample of 168 unrelated male individuals from the Basque Country. Individuals were considered autochthonous if the 8 surnames and birthplace of their grandparents were of Basque origin. The haplotype diversity (99.03%) and discrimination capacity (82.14%) were calculated. The combined polymorphism provides a powerful discrimination tool for routine forensic applications. © 2005 Elsevier B.V. All rights reserved.

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## 1. Introduction

Short tandem repeats on the non-recombining position of the Y-chromosome (Y-STRs) have proven to be of great forensic relevance in special cases where the highly informative autosomal STRs are not applicable and are generally used for male genotyping in sexual

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\* Corresponding author. Tel.: +34 94 6079533; fax: +34 94 6079500.

E-mail address: gobies01@euskalnet.net (O. García).

Table 1

Some 17 Y-STR haplotypes detected in a Basque Country autochthonous population

H21	2	14	11,13	12	28	24	11	13	13	15	12	12	19	15	17	25	12
H29	6	14	11,14	12	28	24	11	13	13	15	12	12	19	16	16	23	12
H42	2	14	11,14	13	29	24	10	13	13	15	12	12	19	16	17	24	12
H45	3	14	11,14	13	29	24	11	13	13	14	12	12	18	16	17	23	12
H52	4	14	11,14	13	29	24	11	13	13	15	12	11	19	15	18	23	13
H55	5	14	11,14	13	29	24	11	13	13	15	12	12	19	15	17	23	12
H57	2	14	11,14	13	29	24	11	13	13	15	12	12	19	15	18	23	13
H58	3	14	11,14	13	29	24	11	13	13	15	12	12	19	16	16	23	12
H75	3	14	11,14	13	30	24	11	13	13	15	12	12	19	16	17	23	13
H81	2	14	11,14	14	30	24	10	13	13	15	12	12	19	16	17	23	12
H85	3	14	11,14	14	30	24	11	13	13	15	12	11	19	15	18	23	11
H94	2	14	11,14	14	31	24	10	13	13	14	12	12	18	15	19	23	11
H102	2	14	11,15	14	30	24	11	13	13	15	12	11	19	15	17	23	12
H106	2	14	12,14	13	29	24	11	13	13	15	12	11	19	15	17	23	12
H107	2	14	12,14	13	29	24	11	13	13	15	12	11	19	15	18	23	12
H111	2	14	12,14	14	30	24	11	13	13	14	12	12	18	15	17	23	11
H115	2	14	12,15	14	30	24	11	13	13	14	12	11	18	15	17	23	11

assault and sibship analyses. However, forensic application of these linked markers requires knowledge of haplotype frequencies from different populations.

In order to create a database representing the Basque Country autochthonous population, the haplotype frequency of the following Y-STRs DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 (Y GATA C4) and Y GATA H4 was established.

## 2. Material and methods

Whole blood was obtained from 168 unrelated Basque autochthonous donors. Individuals were considered autochthonous if the 8 surnames and birthplace of their

Table 2

17 Y-STRs gene and haplotype diversities in Basque Country autochthonous population

	Number of alleles	Gene diversity
DYS389II	6	0.6736
DYS458	6	0.6545
DYS389I	4	0.6170
DYS385	24	0.6169
DYS456	6	0.6155
DYS439	4	0.5855
GATA H4	5	0.5748
DYS448	7	0.5571
DYS437	3	0.4946
DYS391	4	0.4572
DYS390	5	0.4248
DYS635	7	0.3268
DYS19	6	0.3067
DYS392	4	0.2532
DYS393	4	0.2378
DYS438	4	0.2330

Table 3

Different values of HD and DC in the Basque Country population using different panels of markers

Panel of markers	N	Haplotypes	HD	DC
MinHT	168	74	0.9447	0.4405
SWGAM (MinHT plus DYS438 and DYS439)	168	88	0.9689	0.5238
Promega's kit (SWGAM plus DYS437)	168	99	0.9773	0.5893
AB's kit (this study)	168	138	0.9903	0.8214

grandparents were of Basque origin. Genomic DNA was extracted by the standard phenol/chloroform extraction procedure.

PCR amplification was performed using at least 1 ng target DNA in a 25  $\mu$ l final reaction volume according to the manufacturer's recommendations using the AmpFISTR Yfiler PCR Amplification kit (Applied Biosystems, Foster City, CA, USA) that coamplifies 17 Y-STRs. The panel of markers includes the 9-locus European minimal haplotype (minHT) and the markers DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 (Y GATA C4) and Y GATA H4.

Samples were denatured for 5 min at 95 °C and typed on an ABI310 sequencer.

Allele designations were made according to recommendations of the DNA Commission of the International Society for Forensic Genetics [1].

Haplotype and allele frequencies were estimated by gene counting. Gene and haplotype diversities were calculated according to Nei [2] using the Arlequin software, version 2000 [3], and the discrimination capacity was the percentage proportion of the different haplotypes.

### 3. Results and discussion

Table 1 shows some results of the 17 Y-STR haplotype Basque Country autochthonous population database, mainly haplotype shared by several individuals (Haplotype/N/DYS19/DYS385/DYS389I/DYS389II/DYS390/DYS391/DYS392/DYS393/DYS437/DYS438/DYS439/DYS448/DYS456/DYS458/DYS635/Y GATA H4). The most frequent haplotype (3.57%) was found in 6 individuals.

Using this set of Y-STRs markers, a total of 168 unrelated individuals produced 138 different haplotypes of which 121 were singletons.

Gene diversity values of the 17 Y-STR loci in the Basque Country autochthonous population ranged from 0.6736 at DYS389II to 0.2330 at DYS438 (Table 2).

The haplotype diversity (HD) and the discrimination capacity (DC) calculated in this study were 0.9903 and 0.8214, respectively (Table 3).

### References

- [1] DNA recommendations, Report concerning further recommendations of the DNA Commission of the ISFH regarding PCR-based polymorphisms in STR (short tandem repeat) systems, *Int. J. Leg. Med.* 107 (1994) 159–160.
- [2] M. Nei, *Molecular Evolutionary Genetics*, Columbia University Press, New York, USA, 1987.
- [3] S. Schneider, D. Roessli, L. Excoffier, *Arlequin Ver. 2000: A Software for Population Genetics Data Analysis*, Genetics and Biometrics Laboratory, University of Geneva, Switzerland, 2000.